SEPTIN9 gene

septin 9

Normal Function

The *SEPTIN9* gene provides instructions for making a protein called septin-9, which is part of a group of proteins called septins. Septins are involved in a process called cytokinesis, which is the step in cell division when the fluid inside the cell (cytoplasm) divides to form two separate cells. The septin-9 protein also seems to act as a tumor suppressor, which means that it regulates cell growth and keeps cells from dividing too fast or in an uncontrolled way.

The *SEPTIN9* gene seems to be turned on (active) in cells throughout the body. Approximately 15 slightly different versions (isoforms) of the septin-9 protein may be produced from this gene. Some types of cells make certain isoforms, while other cell types produce other isoforms. However, the specific distribution of these isoforms in the body's tissues is not well understood. Septin-9 isoforms interact with other septin proteins to perform some of their functions.

Health Conditions Related to Genetic Changes

Hereditary neuralgic amyotrophy

A few *SEPTIN9* gene mutations have been identified in individuals with hereditary neuralgic amyotrophy, a disorder characterized by episodes of severe pain and muscle wasting (amyotrophy) in the shoulders and arms. The most common mutation results in the replacement of the protein building block (amino acid) arginine with the amino acid tryptophan at position 88 in the septin-9 protein sequence, written as Arg88Trp or R88W. This mutation has appeared in several unrelated families from different parts of the world. Duplication of genetic material within the *SEPTIN9* gene has also been identified in affected individuals.

Changes in the *SEPTIN9* gene may alter the sequence of amino acids in certain septin-9 isoforms in ways that interfere with their function. These mutations may also change the distribution of septin-9 isoforms and their interactions with other septin proteins in some of the body's tissues. This change in the functioning of septin proteins seems to particularly affect the network of nerves controlling movement and sensation in the shoulders and arms (brachial plexus), but the reason for this is unknown.

Because many of the triggers for episodes of hereditary neuralgic amyotrophy also affect the immune system, researchers believe that an autoimmune reaction may be involved in this disorder. However, the relation between *SEPTIN9* gene mutations and immune function is unclear. Autoimmune disorders occur when the immune

system malfunctions and attacks the body's own tissues and organs. An autoimmune attack on the nerves in the brachial plexus likely results in the signs and symptoms of hereditary neuralgic amyotrophy.

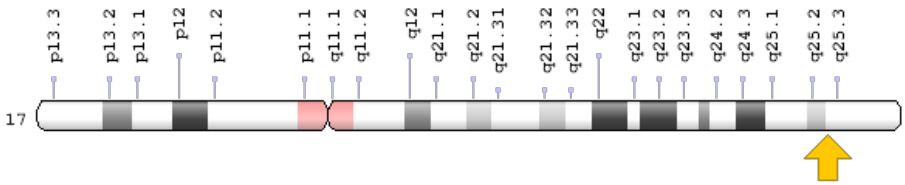
Cancers

Alterations in the activity (expression) of the *SEPTIN9* gene are associated with certain cancers. The altered gene expression may enhance several cancer-related events such as cell division (proliferation), cell movement, and the development of new blood vessels (angiogenesis) that nourish a growing tumor. Increased production of particular isoforms of the septin-9 protein has been associated with breast and prostate cancers. Altered *SEPTIN9* gene expression has also been found in many other cancers, including tumors of the ovary, pancreas, lung, kidney, liver, thyroid and esophagus.

Chromosomal Location

Cytogenetic Location: 17q25.3, which is the long (q) arm of chromosome 17 at position 25.3

Molecular Location: base pairs 77,281,499 to 77,500,596 on chromosome 17 (Homo sapiens Updated Annotation Release 109.20200522, GRCh38.p13) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- AF17q25
- FLJ75490
- KIAA0991
- MLL septin-like fusion
- MSF
- MSF1
- NAPB
- Ov/Br septin
- ovarian/breast septin

- PNUTL4
- SEPT9
- SEPT9_HUMAN
- SeptD1
- septin 9 isoform a
- septin 9 isoform b
- septin 9 isoform c
- septin 9 isoform d
- septin 9 isoform e
- septin 9 isoform f
- septin D1
- SINT1

Additional Information & Resources

Educational Resources

- Molecular Biology of the Cell (fourth edition, 2002): Cytokinesis
  https://www.ncbi.nlm.nih.gov/books/NBK26831/

Clinical Information from GeneReviews

- Hereditary Neuralgic Amyotrophy
  https://www.ncbi.nlm.nih.gov/books/NBK1395

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28SEPT9%5BTIAB%5D%29+OR+%28septin+9%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D

Catalog of Genes and Diseases from OMIM

- SEPTIN 9
  http://omim.org/entry/604061

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/MSFID208.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=SEPTIN9%5Bgene%5D

- HGNC Gene Symbol Report
  https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:7323
- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:10801
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/10801
- UniProt
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